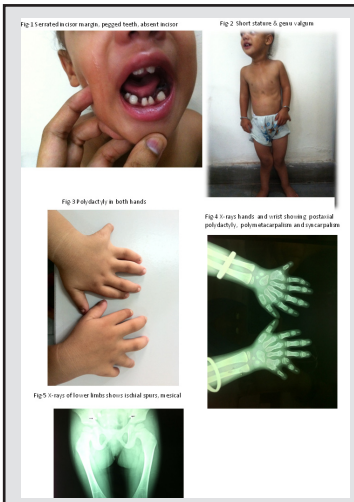
SPOT DIAGNOSIS (IMAGE GALLERY)



GENU VALGUM AND POLYDACTYLY

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A 3 years old girl presented with knock knees and polydactyly. She was first in birth order and single child born to non-consanguineous married parents and had normal milestones. She also had brittle nails. On examination, she had thin hair, slightly elongated face, serrated incisor margins, absent incisor teeth, pegged teeth and transposition of teeth apart from genu valgum and polydactyly. There was no organomegaly. X- ray wrist and hand showing postaxial polydactyly, polymetacarpalism and syncarpalism with shortening of middle phalanges. X-rays of lower limbs shows ischial spurs at acetabulum level, mesical angulation

and shortening of tibia and fibula. X-ray chest showed borderline cardiomegaly with narrow chest and echocardiography revealed large atrial septal defect (ASD).

What is the diagnosis?

Ellis Van Crevald Syndrome (EVC). It is a rare mesenchymal ectodermal dysplasia first described in 1940 by Richard WB Ellis of Edinburg and Simon Van Crevald of Amsterdam now known as EVC syndrome. (1) It is autosomal recessively inherited. (2) EVC is thought to be due to mutation in EVC and EVC – 2 genes located on chromosome 4p16. (3) Child usually has constant finding of polydactyly which is usually bilateral, post-axial and on ulnar side. Polydactyly of feet is present only in 10 percent of patients. (3) Mesomelic shortness of limbs affecting distal segment of limbs is present. Nails are hypoplastic, friable and sometimes absent. Disproportionate dwarfism is there which becomes apparent with subsequent growth. Ischial spurs, genu valgum, narrow chest with poorly developed ribs are present. Oral manifestations are varied including pegged teeth or hypoplastic teeth, accessory labio-gingival frenulum, dental transpositions. Cardiac abnormalities occur in 50-60 percent of cases including AV canal defect, ASD (large) single atrium. (4) One-third of these patients die at early age or infancy from cardio-respiratory problem and those who survive require multidisciplinary approach for treatment i.e. orthopaedic correction of genu valgum, amputation of extra digit, surgical repair of cardiac malformation and dental interventions. Those who survive have normal life span and intelligence in the normal range but final adult height is between 43-60 inches. (5)

References:

1. Shilpy S, Nikhil M, Samir D. Ellis van Creveld syndrome. J Indian Soc Pedod Prev Dent. 2007; 25 Suppl: S5-7.
2. Stoll C, Dott B, Roth MP, Alembik Y. Birth prevalence rates of skeletal dysplasias. Clin Genet 1989; 35: 88-92
3. Kurian K, Shanmugam S, HarshVvardhah T, Gupta S. Chondroectodermal dysplasia (Ellis van Creveld syndrome): A report of three cases with review of literature. Indian J Dent Res 2007; 18: 31-34
4. Baujat G, Le Merrer M. Ellis-van Creveld syndrome. Orphanet J Rare Dis. 2007; 2: 27
5. Chen H, Laufer-Cahana A. Ellis-van Creveld syndrome. Available from: www.emedicine.com/ped/topic660.htm. Cited 10 Jul 2006

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